



## HOGA1 gene

4-hydroxy-2-oxoglutarate aldolase 1

### Normal Function

The *HOGA1* gene provides instructions for making the 4-hydroxy-2-oxoglutarate aldolase (HOGA) enzyme. This enzyme is found in liver and kidney cells, specifically within structures called mitochondria, which are the energy-producing centers in cells. The HOGA enzyme is involved in breaking down a protein building block (amino acid) called hydroxyproline. Specifically, during the breakdown process, HOGA cuts (cleaves) a substance called 4-hydroxy-2-oxoglutarate to produce two smaller substances called pyruvate and glyoxylate. In mitochondria, pyruvate is likely involved in energy production, but the function of glyoxylate is unclear.

### Health Conditions Related to Genetic Changes

#### primary hyperoxaluria

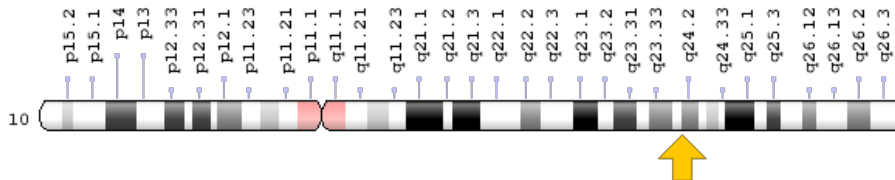
At least 24 mutations in the *HOGA1* gene have been found to cause primary hyperoxaluria type 3. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which begin in early childhood and often result in blood in the urine (hematuria) and urinary tract infections. A specific mutation that alters the way the gene's instructions are used to make the enzyme (written as 700+5G>T) is present in about half of affected individuals. As a result of *HOGA1* gene mutations, the HOGA enzyme cannot break down 4-hydroxy-2-oxoglutarate, which leads to a buildup of this substance in the mitochondria of liver cells.

It is unclear how an accumulation of 4-hydroxy-2-oxoglutarate leads to an overproduction of oxalate in people with primary hyperoxaluria type 3. Some researchers think that the accumulation of 4-hydroxy-2-oxoglutarate interferes with the activity of other enzymes, which lead to the accumulation of substances that get converted into oxalate. Other researchers think that excess 4-hydroxy-2-oxoglutarate in mitochondria may leak out into liver cells. Enzymes within these cells would then convert 4-hydroxy-2-oxoglutarate to glyoxylate, and then convert glyoxylate into oxalate. In individuals with primary hyperoxaluria type 3, the oxalate is filtered through the kidneys and is either excreted in urine as a waste product or combines with calcium to form calcium oxalate, a hard compound that is the main component of kidney and bladder stones.

## Chromosomal Location

Cytogenetic Location: 10q24.2, which is the long (q) arm of chromosome 10 at position 24.2

Molecular Location: base pairs 97,584,345 to 97,612,802 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DHGPS2
- DHGPSL
- dihydrodipicolinate synthase-like, mitochondrial
- dihydrodipicolinate synthetase homolog 2

## Additional Information & Resources

### GeneReviews

- Primary Hyperoxaluria Type 3  
<https://www.ncbi.nlm.nih.gov/books/NBK316514>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HOGA1%29+OR+%284-hydroxy-2-oxoglutarate+aldolase+1%29+OR+%28primary+hyperoxaluria+type+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- 4-HYDROXY-2-OXOGLUTARATE ALDOLASE 1  
<http://omim.org/entry/613597>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=HOGA1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=25155](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25155)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/112817>
- UniProt  
<http://www.uniprot.org/uniprot/Q86XE5>

## **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22391140>

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